

FIG. 1

a L-periaxin

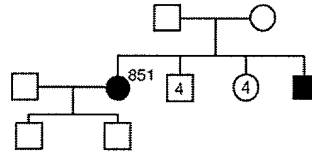
L-peraxin		PDZ Domain					
Human	1	MEARSRAEELRRAELVEIIVETAQTGVSGINVAGGGKEGIFVRELREDS	PAARSLSLQEGDQLLSARVFFENFKYEDALRLQLCAEPYKVSFCLK	97			
Murine	1F.....K.....	97			
Rat	1F.....K.....	97			
		NLS1	NLS2	NLS3			
Human	RTVPTGDALALRP	GTSGYEIKGPRAKVAKLNIQSLSPVKKK	KHVP	GALGVPADLAPVDVEFSFPKESRLRRGLKAEAVKGPVPAAPARRRLQLPRLRVRE	197		
MurineM.....A.....T.....T.....	197		
RatM.....A.....T.....T.....	197		
Human	VAEEAQAARLAAAAPP	PRKAKVEAEVAGARFTAPQVELVGRPLPGA	EVGPVQVSAKAAPSAEAAAGFALHLP	TLGLGAPAPPAVEAPAVGIQVPQVEL	297		
MurineV.....M.....A.....A.....T.....G.....I.....S.....V.....GT.....T.....S.....P.....T.....	297		
RatV.....M.....S.....T.....G.....I.....S.....K.....V.....GT.....T.....S.....A.....P.....T.....	297		
Human	PALPSLPTLPTL	PCL	ETREGAVSVVPTLDVAAPT	VGVDLALPGA	VEARGEAEVALKMPRL5FPRFGARAKEVAEAKVAKVSP	PEARVKGPRLRMPTFG	397
MurineT.....D.....Q.....AV.....K.....F.....SM.....Q.....V.....I.....G.....AT.....V.....G.....KA.....	397	
RatT.....D.....Q.....AV.....K.....F.....SM.....Q.....V.....V.....G.....AT.....V.....G.....KA.....	397	
			Repeat Domain				
Human	LSLLEPRPAPE	-VVESKLKLP	TIKHP	SLGIGVSGPEVKVPKGEVKLPK	PAEALPEVRLPEVELPKVSEMKLPKVP	EMAVPEVRLPEVEL	496
MurineS.....G.....A.....A.....L.....F.....A.....A.....T.....V.....I.....D.....Q.....Q.....M.....D.....I.....V.....D.....Q.....	497
RatS.....G.....VAA.....L.....F.....S.....A.....V.....I.....	453
Human	PKVSEMKLPKVP	EMAVPEVRLPEVQLLKVSEMKLPKVP	EMAVPEVRLPEVQLPKVSEMKLP	EVSEVAVPEVRLPEVQLPKVPEMKVP	EMKLPKVP	EMKLP	596
MurineP.....V.....D.....H.....D.....P.....K.....P.....M.....D.....H.....D.....AV.....	576
RatAP.....AAI.....D.....Q.....P.....MSD.....D.....H.....K.....P.....V.....MKLPKT.....MAV.....D.....H.....DIQLP.....DKLP.....V.....	545
Human	EMKLPEVQLPKV	-----PEMAVPDVHLPEVQLPKVPEMKLP	EMKLPEVQLPKVPEMAVPDVHLPEVQLPKVPEMKLP	KMP	EMAVPEVRLPEVQLPKV	689	
Murine	DVR.....SEVKLPKM.....L.....MS.....M.....R.....S.....L.....	665	
RatAV.....D.....H.....DIQLPKVPEMKLPDKLPKVP.....MAV.....D.....IPEVQLPVS.....L.....D.....R.....L.....MS.....V.....I.....D.....D.....	645	
Human	SEMKLPKVP	EMAVPDVHLPEVQLPKVCEMKVPDMKLPEIKLPKVP	EMAVPDVHLPEVQLPKVSEIRLPEMQVPKVPDVHLPKA	PEVKLPRAPEVQLKATK	788		
MurineI.....L.....E.....D.....P.....L.....D.....SQ.....E.....Q.....M.....SKV.....R.....SAG	740	
RatL.....TM.....IR.....T.....P.....I.....Q.....KV.....A.....R.....SAG	730	
Human	AEQAEGHEFGKMPKHTMPKLGRAE	SPSRGKPG	EAGAEVSGKLVTLPC	LQPEVDGEA-HVGVPSLTLPSVELDLPGALGLQGVPA	AKMGKGERAE	GPEV	887
MurineK.....T.....S.....L.....V.....KV.....SI.....PD.....MT.....GT.....S.....S.....QE.....VP.....V.....KP.....R.....	835
RatKT.....S.....L.....V.....KV.....PD.....LI.....GT.....VAR.....S.....QE.....VS.....V.....KP.....R.....	825
Human	AAGVREVGFRVPSVEI	VTQPLPAVEIEEGRLEMTETKVPSSKFSLPKFGLSGPKVAKAEAG	GRATKLVSKFAISLPKARVGAEAEAKGAGEAGLLP	987			
MurineV.....G.....TV.....V.....KEQ.....V.....M.....MS.....M.....R.....T.....A.....T.....	935	
RatV.....TG.....A.....N.....T.....VKKEQ.....V.....M.....T.....AV.....V.....P.....R.....A.....TD.....D.....	925	
Human	ALDLSIPQLSLDAHLPSGKVEVAGADLKFKGPRFALPKF	GVGRDTEAAELVPGVAELGKGGWGDGRVKMPKLMPSFGLARGKAE	EVQGDRA	SPGEKA	1087		
MurineQ.....S.....P.....SS.....K.....S.....DV.....A.....E.....K.....S.....T.....DG.....V.....L	1033	
RatQ.....S.....P.....ES.....P.....S.....AK.....S.....DV.....A.....E.....K.....S.....T.....DG.....V.....L	1025	
			Acidic Domain				
Human	ESTAVQLKIPEVELVTLGAQE	EGRAGAVSGHQLSGKLVSTARQVTECHDAGLRHPP	LGESLPQVELTGFG	EAGTPGQQAQSTVPSAEGTAGYRVQV	1187		
MurineAI.....G.....K.....P.....TEK.....T.....VKP.....Q.....TC.....A.....QESVQVST.....AS.....ETV.....S.....	1123	
RatAI.....G.....P.....TEK.....T.....VKP.....Q.....T.....QEGAQVSS.....AS.....ETA.....V.....S.....I.....	1115	
Human	PQVTLSP	GAQVAGGELLVGGGVFKMPTVTVPQLELDVGLSRE	AQAGEAATGEGGLRLKLP	TLGARAVGGEGAEQPPGAERTFCLSLPDVELSP	SGGN	1287	
MurineM.....E.....T.....D.....I.....GH.....K.....S.....IK.....GTGSR.....V.....P.....G.....E.....Q.....H.....TSPVSS	1221	
RatM.....E.....T.....D.....I.....GH.....T.....KS.....K.....G.....GGK.....A.....S.....E.....QH.....HI.....TSPVSS	1213	
Human	HAEYQVAEGEGEAGHKLKVR	LPFRGLVRAKEAGEE	EKAQSPKRLPRVGF	SQSEHMTGEGSPSP	EEEEEEEEEGSGEGASGRGRVRLPRVGLAAPS	1387	
MurineV.....D.....DG.....L.....AK.....I.....V.....S.....S.....S.....S.....S.....S.....	1317	
RatV.....D.....DG.....L.....A.....I.....T.....V.....S.....S.....S.....S.....S.....S.....	1309	
Human	KASRGQEGDAAPKSPVREKSPKFR	PRVSLSPKARSGSDQEEGGLRVR	LPVSGFSETG-----APGPARMEGAQAAAV*	1461			
MurineV.....K.....TS.....G.....R.....F.....TA.....VP.....T.....I.....T.....I.....	1391	
RatV.....K.....TS.....G.....R.....F.....TA.....VP.....T.....I.....T.....I.....	1391	

b S-periaxin

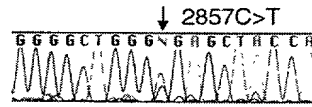
			PDZ Domain	
Human	1	MEARSRAEELRRAELVEIIVETAQTGVSGINVAGGGKEGIFVRELREDS	PAARSLSLQEGDQLLSARVFFENFKYEDALRLQLCAEPYKVSFCLK	97
Murine	1F.....K.....	97
Rat	1F.....K.....	97
Human	RTVPTGDALRP	GTSGYEIKGPRAKVAKLVRVLS	PAPALDCPSDPVSA-P*	147
MurineM.....V.....VQ.....S.....R.....A.....A.....	148
RatM.....A.....	147

FIG. 2

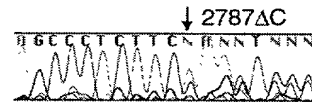
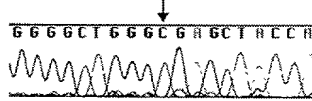
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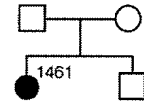
Patient 851



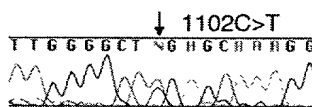
Wild type



HOU 579



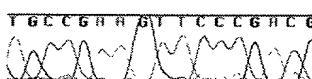
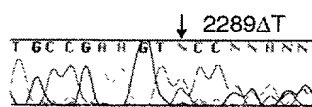
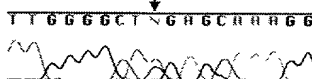
Patient 1461



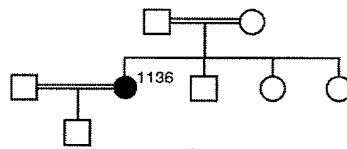
Father



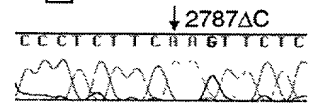
Mother



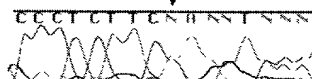
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Patient 1136



Father



Mother

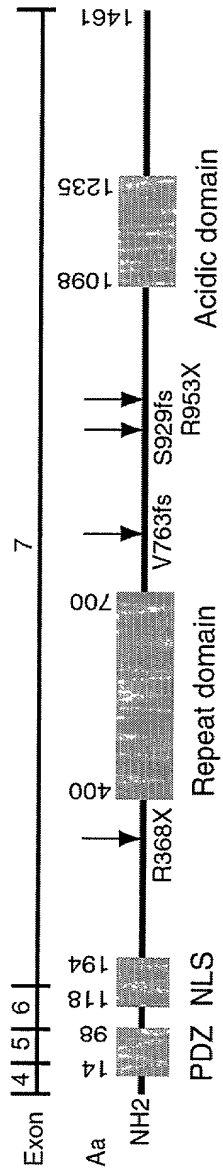


Wild type



FIG. 3

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Patient	Phenotype	Pathogenic Allele 1	Frequency	Pathogenic Allele 2	Frequency
851	DSN	2857C>T R953X	0/190	2787ΔC S929fsX957	0/190
1461	DSN	1102C>T R368X	0/178	2289ΔT V763fsX774	0/176
1136	DSN	2787ΔC S929fsX957	0/190	2787ΔC S929fsX957	0/190

FIG. 4

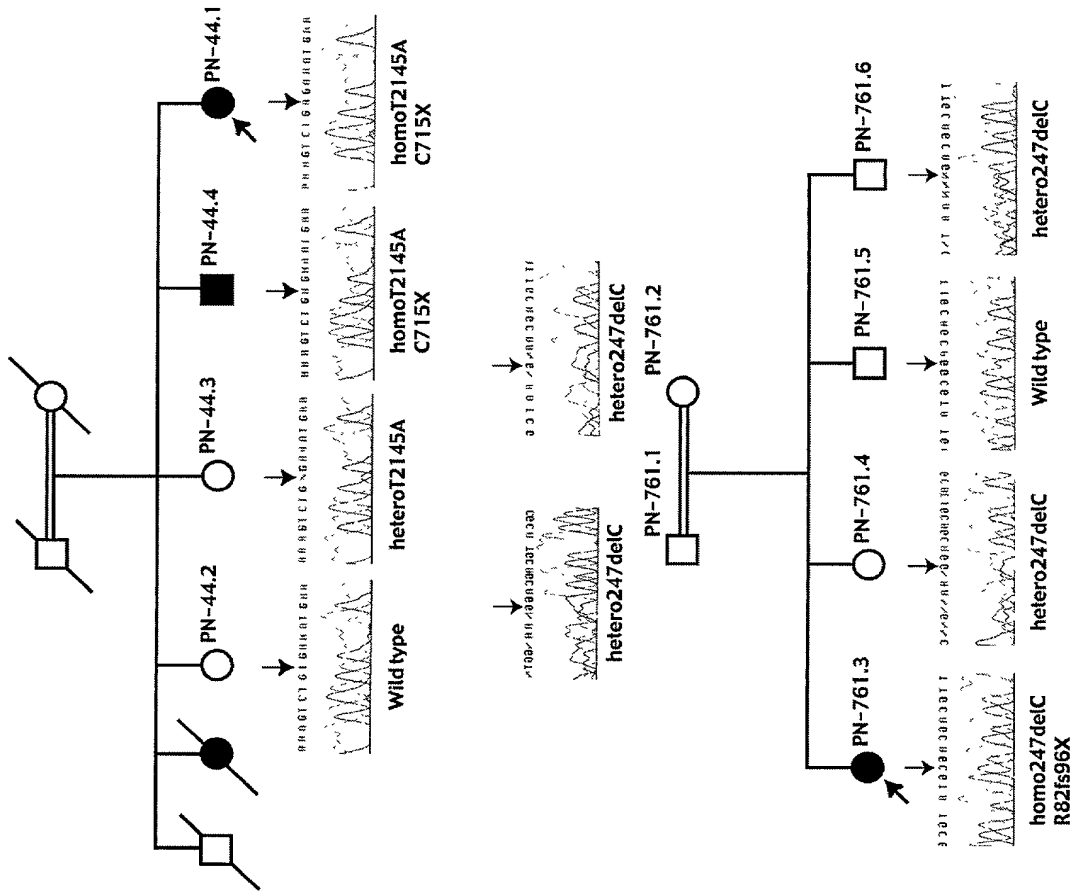


FIG. 5

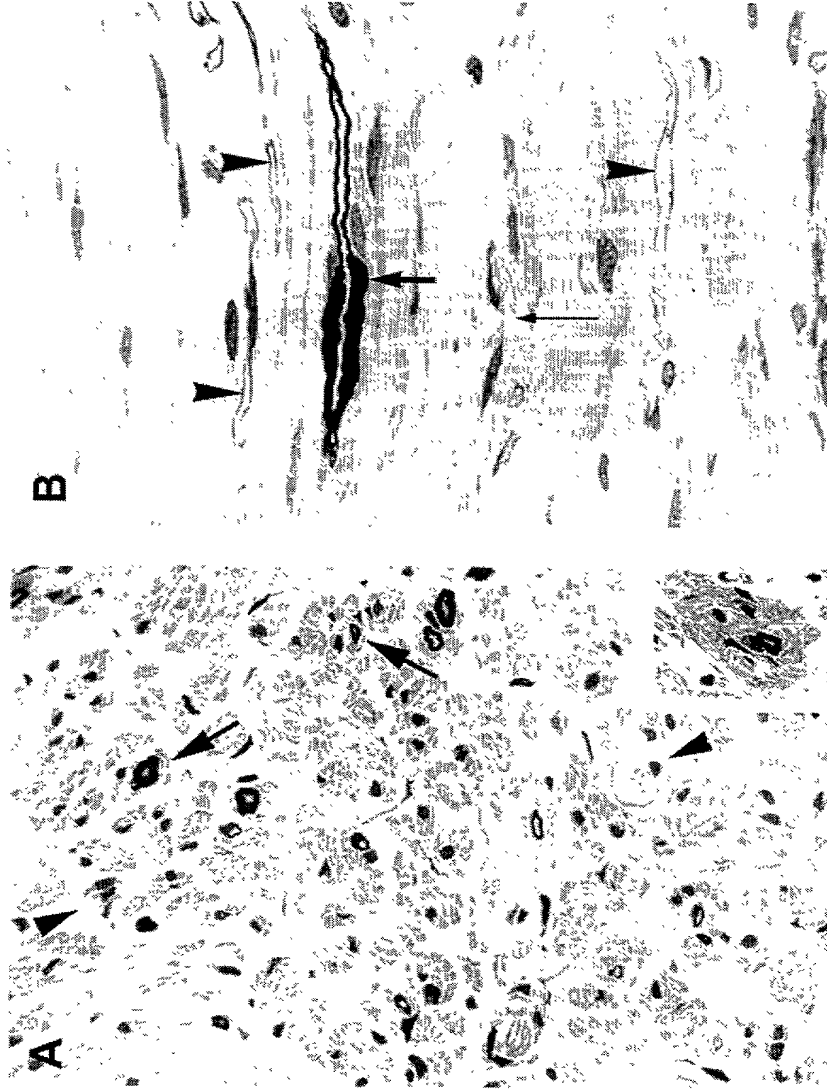


FIG. 6

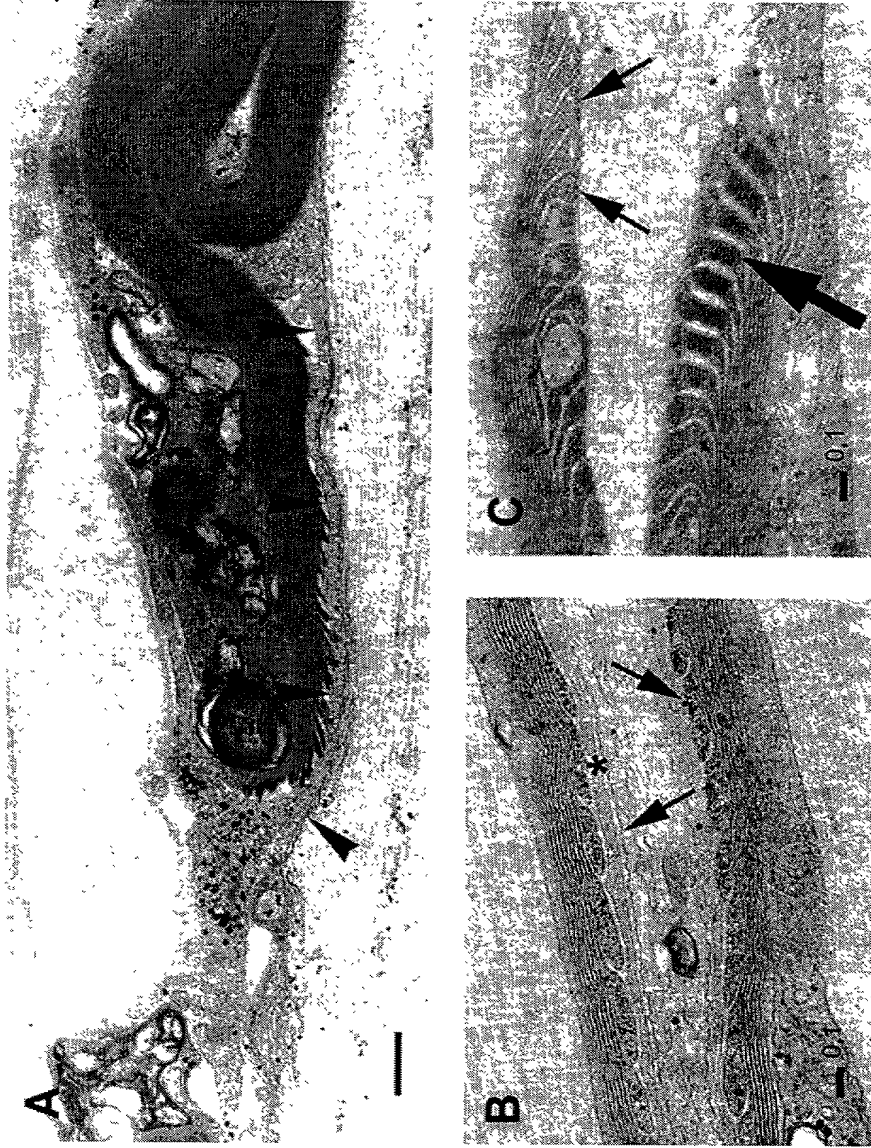


FIG. 7

Figure 8 shows the results of the experiment. The figure is divided into four panels, A, B, C, and D. Each panel shows a series of images of a cell, with the cell's position and orientation indicated by a small white arrow. The images are arranged in a grid, with the cell's position and orientation indicated by a small white arrow. The images are arranged in a grid, with the cell's position and orientation indicated by a small white arrow.

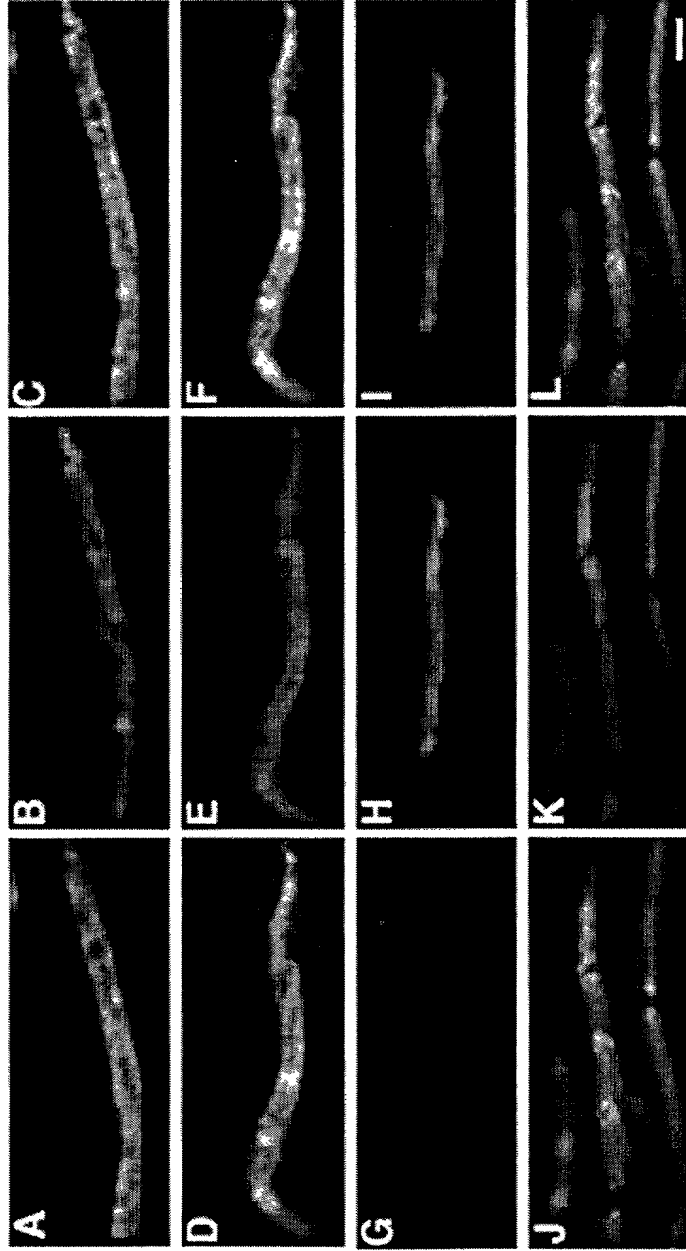
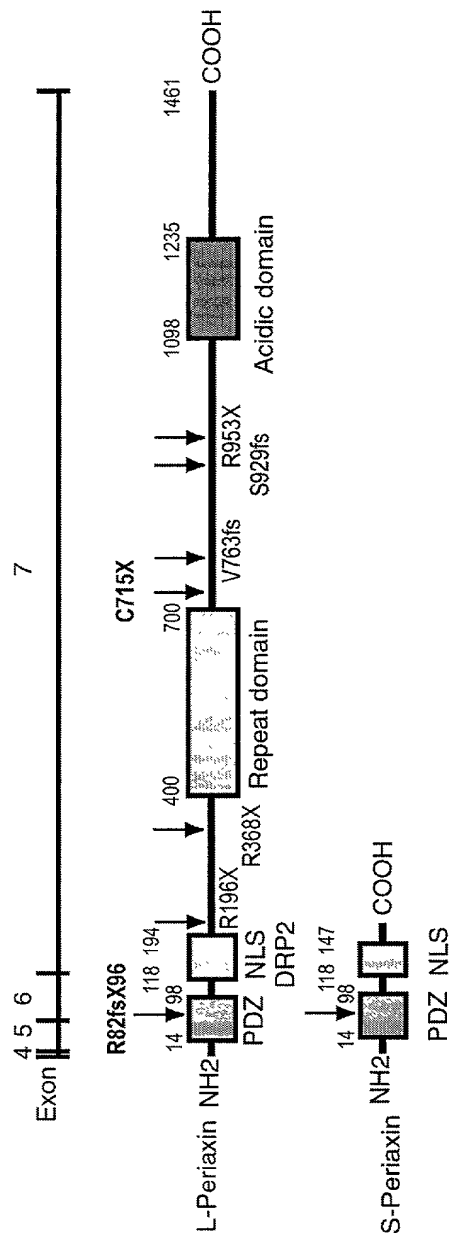


FIG. 8



Patient	Phenotype	Allele 1		Allele 2	
		Pathogenic	Frequency	Pathogenic	Frequency
PN44.1	CMT	C715X	0/180	C715X	0/180
PN44.4	CMT	C715X	0/180	C715X	0/180
PN761.3	CHN	R82fsX96	0/190	R82fsX96	0/190
851	DSN	R953X	0/190	S929fsX957	0/190
1461	DSN	R368X	0/178	V763fsX774	0/176
1136	DSN	S929fsX957	0/190	S929fsX957	0/190
CMT4F	DSN	R196X		R196X	

FIG. 9